CASE REPORT

Pseudohypoparathyroidism – A Clinical Rarity

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Abstract

Pseudohypoparathyroidism is a metabolic disease caused by an inability of the body to respond appropriately to parathyroid hormone (PTH), the principal hormone that regulates calcium and phosphorous levels in the body. Due to the disturbance in the peripheral action of parathormone there is a high serum level of parathormone characterized by hypocalcaemia. Parathormone deficiency has an effect on the calcification of the dental tissues and to a lesser extent on bone matrix production, but the mechanism is obscure.
This case report emphasizes the pertinent oral findings which may provide the diagnostic aids for the disease. Therefore, the fundamental difference between hypoparathyroidism and pseudohypoparathyroidism is that PTH is absent in hypoparathyroidism, but the body will respond to it; and PTH is plentiful in pseudohypoparathyroidism, but the body cannot respond to it.

KEYWORDS: pseudohypoparathyroidism, hypoparathyroidism

Introduction

A few rare metabolic disorders and genetic diseases remain unrecognized by the oral specialists and evade the common diagnostic perceptions. Following our experience, here we report one such metabolic disorder. Pseudohypoparathyroidism (PHP) is a rare disease of hypocalcemia syndrome and was first described by Albright and co-workers in 1942 as Albright's Hereditary Osteodystrophy. The disease expresses clinical and biochemical features similar to hypoparathyroidism but fails to respond to parathormone by normal phosphorous diuresis. (Ellsworth-Howard test)[1]

Pseudohypoparathyroidism (PHP) is a heterogeneous group of disorders characterized by hypocalcemia, hyperphosphatemia, increased serum concentration of parathyroid hormone (PTH), and insensitivity to the biological activity of PTH.[2] It may be considered an inborn error of metabolism.[3] Pseudohypoparathyroidism is characterized by an inability of the body to respond appropriately to parathyroid hormone (PTH), the principal hormone that regulates calcium and phosphorous levels in the body.

The syndrome appears to be inherited as an X-linked dominant trait.[4] The most common presenting symptom is tetany or convulsion, which may be misdiagnosed as epilepsy, especially in infants.

The dental manifestations are characterized by enamel hypoplasia, retarded root development with short blunted roots, larger root canals and distorted and thick dentin at the apex. The genetic defect responsible for the disease is amputations of guanine nucleotide-binding a-subunit gene (GNAS), the gene encoding the a-subunit of the stimulatory GTP binding protein.[5]

CASE REPORT

A female patient, aged 11 years reported to the Dept. of Oral Medicine and Radiology with the chief complaint of delayed eruption of her permanent teeth. The patient's mother elicited the history of non-eruption of her permanent teeth and a history of epileptic attack around 2 months back for which she was hospitalized. The patient was apparently normal upon presentation but had the attention deficit disorder with shorts periods of hyperactivity followed by periods of normal activity. The patient's mother gave the history of febrile convulsions during the first 15 days of her infancy with the history of similar absence seizures in the past.

The EEG showed mild abnormality in the form of background slowing which was suggestive of cortical dysfunction or post-ictal phenomenon. She was admitted to the hospital as her serum calcium was lower than normal. The phosphorus and magnesium levels were in the normal limits. The patient's parent also gave the history of cataract in both of her eyes with surgeries done in the left and right eye for the insertion of bifocal lens.

Physical examination demonstrated a healthy appearing shy girl. She was short statured, had delayed developmental milestones. Her height was 109 cms and weighed 19 kilograms. She suffered from early cataracts with bilateral lens opacity for which she had been operated upon and showed slight mental retardation. A history of consanguineous marriage (First degree) was given by the parents. Clinical dental examination revealed enamel hypoplasia in the form of rough and pitted texture and dull yellow to brown color teeth in the deciduous dentition and non eruption of permanent teeth. The gingival mucosa was healthy with presence of deposits around the teeth.(Fig.1a,b)

The radiographs revealed incomplete root formation with respect to the upper and lower central incisors with wide pulp chambers. Upon panoramic examination, the OPG revealed multiple unerupted permanent teeth with widenend pulp chambers and retarded root development. (Fig.2) The lateral skull and P.A. Views were made. The calvaria were thickened and showed no abnormal calcifications in the skull region. (Fig.3)

DISCUSSION

Pseudohypoparathyroidism (PsHP) is a metabolic disease caused by a disturbance in the peripheral action of parathormone.[6] It is characterized by an inability of the body to respond appropriately to parathyroid hormone (PTH), the principal hormone that regulates calcium and phosphorous levels in the body.

Clinically the disease is similar to idiopathic hypoparathyroidism. Idiopathic hypoparathyroidism is a rare endocrinopathy whose origin is unknown, and which is characterized by a deficiency of parathyroid hormone causing low serum calcium and high serum phosphorus concentrations. Disorders of ectodermal tissues are common in this pathology, and enamel hypoplasia is the most frequently found dental pathology.[7]

Characteristically the patients are dwarfed in stature, thick set, and obese, having round faces and greasy skin. There is considerable reduction in the length of the 3rd and 4th fingers and toes and dimpling of the corresponding knuckles when the fist is clenched. Mentally these patients are slightly retarded. Radiographs show that the 3rd and 4th metacarpals and metatarsals are short. Epiphysial closure is in advance of age, and there are patches of soft tissue calcification and ectopic ossification, usually in the thighs. Radiating striae of calcification of the basal ganglia are also described as a frequent finding.

The clinical findings in these cases of pseudohypoparathyroidism are similar to those reported in cases of idiopathic hypoparathyroidism, and may be summarized as follows.[6]

(1) Teeth dull white in colour with hypoplastic pitting.

(2) Crowns are small and the roots are often short with blunt ends.

(3) The enamel is thin and the pulp chambers large, often nearly occluded by calcified deposits.

(4) In some cases there may be many unerupted teeth.

(5) A full complement of teeth is not always developed, premolars being the teeth most usually missing.

(6) The teeth are lost early due to caries.

(7) The jaws are short and wide in all cases.

In our case, we took a prosthodontic consultation and decided upon the fabrication of removable partial dentures which may aid in the eruption of permanent teeth. (Fig.4a,b)

CONCLUSION

A patient with a systemic disorder, such as PsHP, which is a rare metabolic disorder, requires careful anamnesis, and thorough clinical and radiographic examinations. The clinician must be alert to oral abnormalities such as enamel hypoplasia, widened pulp chambers, shortened roots, hypodontia, and delay or cessation of dental growth and development, which suggest a systemic disorder. The main goal of therapy in PsHP patients is to restore serum calcium and phosphorus. A multidisciplinary approach is key to the correct diagnostic and treatment choice aiming to improve the patient's health.

Fig. 1.a,b – Frontal Extra Oral And Intra Oral View

Fig. 2 – OPG View

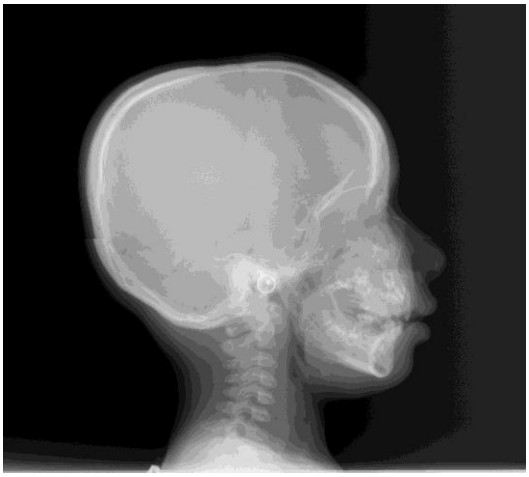

Fig. 3 – Lateral Skull View

Fig. 4.a,b – Upper and Lower Partial Dentures

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